

Alternative cascade-testing protocols for identifying and managing patients with familial hypercholesterolaemia: systematic reviews, qualitative study and cost-effectiveness analysis

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Plain language summary

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Plain language summary

Familial hypercholesterolaemia is an inherited condition that causes raised cholesterol levels from birth and increases risk of heart disease if left untreated. After someone in a family is found to have familial hypercholesterolaemia (called an index case), their close relatives need to be contacted and checked to see if they have familial hypercholesterolaemia, using genetic or cholesterol testing. This is called 'cascade testing'. We planned to find the most cost-effective and acceptable way to do this.

The relatives could be contacted for testing by the index case (indirect approach), by a health-care professional (direct approach) or by a combination of both approaches. We found, based on looking at hospital records, that more relatives were tested if health-care professionals directly contacted relatives. In previous studies, slightly more relatives were tested for familial hypercholesterolaemia with a combination approach. Interviews with patients also suggested that the direct approach was the most effective, but the most acceptable and successful approach depends on family relationships: using one approach for some families and using both for other families.

Furthermore, by looking at the health-care records of large numbers of patients, we confirmed that people with a recorded diagnosis of familial hypercholesterolaemia in general practice records have a much higher risk of heart disease than the general population, and this was especially so for those with previous heart disease and/or raised cholesterol levels when diagnosed. However, one-quarter of new patients with familial hypercholesterolaemia recorded in their records were not treated within 2 years, with less than one-third reaching recommended cholesterol levels.

We used what we had learned to help us estimate the most cost-effective way to do cascade testing. This showed that if the health service directly contact all relatives simultaneously for further assessment, rather than the current approach whereby close (first-degree) relatives are contacted first, this was cost-effective and good value for money.

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This report

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